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Doctor of Physical Therapy Program Case Reports

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## Use of Aquatic Based Physical Therapy in Treating an Adolescent with Severe Type II Spinal Muscle Atrophy: A Case Report

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### Abstract

**Background:** Spinal muscle atrophy (SMA) is a genetic disorder of the motor neurons resulting in muscle atrophy and weakness of varying degrees based on severity and type. Although there is current research regarding forms of gene therapy and medications to treat this disease, there is limited research on the most effective physical therapy interventions, especially for more severe types.

**Purpose:** The purpose of this case study is to discuss the therapeutic benefits of aquatic physical therapy in an individual with compromised neuromuscular control, even when gross motor outcome measures did not reflect measureable change. **Case Description:** The patient was a 17 year old male diagnosed with severe type II spinal muscle atrophy. His primary goals for therapy were to address back, wrist and neck pain. As a child, he never reached any gross motor developmental milestones. He utilized a power-wheelchair for mobility and required maximal assistance for all transfers. **Intervention:** The patient was seen for one land-based session and one aquatic-based session each week over the course of two years. **Outcome Measures:** Manual muscle testing and a SMA specific outcomes assessment did not indicate any measurable change in function. However, the PedsQL may be indicated to demonstrate improved quality of life. **Discussion:** The use of aquatic physical therapy for individuals with degenerative neuromuscular disorders can provide an environment of safety that optimizes their ability to utilize volitional muscular control even in the absence of measureable gross motor gains. Therapeutic benefits of aquatic physical therapy interventions in cases like this may be measured best by use of quality of life measures, such as the PedsQL.

## Introduction and Background

Spinal muscle atrophy is defined as a genetic disorder of the motor neurons in the anterior horns of the spinal cord resulting in degeneration of the spinal cord alpha motor neurons. The disease is identified genetically by deletions of the SMN-1 gene. Clinically, these patients will present with muscle atrophy, weakness, loss of reflexes, tongue fasciculation, tremor, and denervation on EMG. Several types of SMA are recognized and vary in severity. Type I is the most severe, and characterized by an onset before 6 months of age when they do not achieve the milestone of independent sitting. Infants with type I SMA appear normal in tone and strength at birth, but quickly show weakness, respiratory distress, poor feeding, and low tone, as well as a bell shaped chest due to poor chest wall expansion linked to poor diaphragmatic function. Type II SMA is less severe, as these children experience onset at approximately 6 to 18 months of age. These individuals will typically achieve the ability to sit independently but do not achieve the gross motor milestone of walking. Proximal weakness as well as greater weakness in the lower extremities compared to upper extremities is common. Reflexes are absent, and scoliosis is common. Survival into adulthood is not abnormal, but increased care for swallowing and respiration is often at a greater need over time. A diagnosis of type III SMA would describe an individual with the diagnosis who has been able to walk at some point in his or her lifetime. The onset of type III is generally at 18 months of age or older. Clinically, these individuals may report frequent falls, difficulty climbing steps and proximal weakness. More recently, type 0 and type IV have also been identified. Type 0 is clinically defined as prenatal onset, and type IV is a milder form that has a later onset.<sup>1</sup>

A recent article published in October 2017 indicates there are a variety of gene therapies available as therapeutic interventions for the disease and that children are being diagnosed sooner allowing for more immediate treatment.<sup>2</sup> The current consensus statement for standard of care in SMA, recommends multidisciplinary care, including physical therapy.<sup>2</sup> However, this statement focuses primarily on fitting assistive devices, with little mention of exercise as a viable component of physical therapy in this population.<sup>3</sup> Physical therapy is currently being provided for a high number of patients with SMA. One study found that only 86 of 105 patients with SMA reported receiving physical therapy services.<sup>4</sup> Of those 86, 62% received therapy in a neuromuscular clinic, 38% at school, 34% at home, and 13% in an outpatient clinic. Greater frequency of PT services received was associated with younger age and inability to walk, but not SMA type. However, this study was unable to assess the impact of PT services on progression of disease or prognosis.<sup>4</sup>

There is limited research supporting the use of physical therapy for milder forms of SMA.. One study examined how a combination of water-based and land-based physical therapy would affect muscle strength testing and “quality of daily living” in a group of patients diagnosed with either type II or type III SMA. This study found that while no significant strength gains were noted with manual muscle testing, function and quality of life was improved using the Barthel Ladder Scale.<sup>1</sup> Additionally, a case study reported that a child with type III SMA demonstrated improved gross motor skill function following the combination of a land-based and aquatic based program..<sup>5</sup> However, there is minimal research evaluating physical therapy interventions in patients with more severe forms of SMA.

Thus, the purpose of this case study is to highlight the benefits of utilizing aquatic based therapy interventions over a two-year time period for a child with a severe case of type II SMA. This case study will emphasize the significance of aquatic based therapy to maximize the patient’s ability to progress with volitional muscular control in an environment of decreased gravity and safety for the patient. An additional purpose of this case study is to provide a patient scenario in which physical therapy services were beneficial to the patient’s quality of life, even in the absence of clinically significant or rapid change in measurable objective gross motor findings or functional mobility. This patient provides a good example of the challenges but potential benefit and positive outcomes of physical therapy interventions to treat a progressive neuromuscular disease.

## Case Description

This patient was a 17 year old Caucasian-Hispanic male with the medical diagnosis of spinal muscle atrophy. He attended public high school, lived with his parents, and was an only child in a middle class neighborhood. His initial diagnosis as an infant was type I SMA because he never achieved independent sitting as an infant. However, he is not and never was on a ventilator, which makes his true diagnosis a severe form of type II SMA. His only medical comorbidity includes mild respiratory insufficiency due to weakened respiratory musculature. The patient and his mother reported no other significant birth history or complications. The patient's other past medical history includes a hairline fracture on his left humerus in 2013 that occurred as an accident when a caregiver was performing a dependent transfer with him. He also had a spinal growing-rod insertion procedure in 2016 from T1-S5 to correct scoliotic curvature with the development of an infection of the lumbar incision after surgery. He also reported having been hospitalized fairly regularly throughout his childhood and into his first two years of high school for pneumonia. He currently utilizes a power wheelchair with a Jay 2 seat and back and is dependent for all transfers. He has some volitional control in his left hand to drive his power chair, but otherwise has very minimal upper extremity control. The patient utilized a g-tube and feeding pump, as well as a suction machine due to difficulty swallowing both food and saliva. For medications, he was taking an anti-secretion medication to reduce salivation and need for suctioning, as well as pain medications including Tylenol as needed and codeine each day before and after school. The patient had school based physical therapy once per week from kindergarten until 7th grade. From 7th grade until sophomore year of high school when he began his current episode of care, he did not have any physical therapy. His current episode of care involves physical therapy sessions twice each week for 60 minutes each. One session is land based and the other is aquatic based. The patient verbalized understanding of his disease being progressive, so his major goals for PT were to gain relief from back, wrist and neck pain.

### Examination and Evaluation:

The initial evaluation was completed by another therapist who saw the patient at the beginning of his care nearly 2 years ago at the same facility. This therapist took initial manual muscle testing (MMT) measurements, as well as hip and knee flexion active range of motion (AROM) measurements (see Tables 1 and 2). Objective observations about musculoskeletal deformities were also noted, but no other outcome measures were utilized at the initial evaluation. Other objective observations included scoliotic curvature and right windswept deformity. No initial pain rating or quality of life outcome measures were assessed at this time.

**Table 2.** Initial Evaluation Active Range of Motion (AROM)

	Right	Left
Hip Flexion	45*	60*
Knee Flexion	120*	90*

**Table 1.** Initial Evaluation Manual Muscle Testing (MMT)

	Right	Left
<b>Hip</b>		
Flexion	2-	2-
Extension	2	2
Internal Rotation	0	0
Lateral Rotation	0	0
Abduction	2-	0
Adduction	2-	2-
<b>Knee</b>		
Flexion	2-	2-
Extension	1	2-
<b>Ankle</b>		
Dorsiflexion	0	1
Plantarflexion	2	2
Inversion	0	0
Eversion	1	1
<b>Great Toe</b>		
Flexion	2-	2-
Extension	2-	2-

## Interventions:

Over the past two years at this outpatient pediatric clinic, treatment included two 60 minutes physical therapy sessions per week, with one being land based and the other being aquatic. Land based therapy sessions would begin with a dependent transfer of the patient from his power chair onto the high-low treatment table. Interventions included passive range of motion (PROM) and stretching in supine of the bilateral upper and lower extremities, as well as a combination of active assisted range of motion (AAROM), active range of motion (AROM) and isometric activation of major lower extremity muscle groups. Treatments would also include working on maintaining and tolerating sitting balance on the edge of the treatment table, increased independence with head control while in sitting, and rolling. The patient was dependent with all position change transfers and all functional mobility work required maximal assistance for safety.

Aquatic based therapy sessions would begin with a dependent transfer into the pool from his power chair utilizing a pool lift and physical assist from the therapist. The therapy pool in this clinic was approximately 10"x12", 4" deep, and kept between 89-92 degrees F. The sessions would typically begin with PROM and stretching in the pool with maximal support from the physical therapist. Then the patient would work on floating on his back and tucking his chin to keep his head above water for improved head control. We would time the patient to see how long he could maintain independent support to track progress. This was done to work on head control. The patient would also work on swimming and walking without physical assist of the physical therapist. He would utilize a neck float for head support during these tasks for improved volitional activation of the limbs and safety. With assistance from the physical therapist, his arms were placed behind his back. He was able to internally and externally rotate at the shoulders and flex and extend at the hip to move himself across the pool. For walking, 1.5lb ankle weights were placed on his ankles to allow for foot contact on the bottom of the pool and the patient was able to initiate small steps with utilizing activation of his hip flexors.

## Outcomes

As the current therapist, I was able to see the patient for 9 weeks, nearly 2 years into his care. The outcomes reported here reflect his current assessments at the end of these 9 weeks. I asked him to describe the percent change he had experienced in pain since beginning with physical therapy at our facility 2 years ago. He reported an 85% decreased in his back, neck and wrist pain. He had also reduced codeine intake to 1 dose per day as compared to 2 doses per day prior to the start of physical therapy. This meant that he did not need to leave class during the day to take pain medications and was better able to focus in class due to decreased pain. In addition, he mentioned that since beginning physical therapy two years ago, he had not been hospitalized for pneumonia. He also stated, "I'm doing things I never thought I'd be able to do" indicating psychological benefits and improved quality of life perception due to increased participation and skills completed within the aquatic environment.

From the clinician's perspective, the patient was able to move more independently in the water and demonstrated greater active range of motion in the water over time. However, these changes were identified by clinical

**Table 3.** Current Status  
Manual Muscle Testing (MMT)

	Right	Left
<b>Hip</b>		
Flexion	2-	2-
Extension	2	2
Internal Rotation	0	0
Lateral Rotation	0	0
Abduction	2-	0
Adduction	2-	2-
<b>Knee</b>		
Flexion	2-	2-
Extension	1	2-
<b>Ankle</b>		
Dorsiflexion	0	1
Plantarflexion	2	2
Inversion	0	0
Eversion	1	1
<b>Great Toe</b>		
Flexion	2-	2-
Extension	2-	2-

observation, with no formal outcome measures utilized to detect this change. The patient agreed that he felt that he was able to move better and more fully in the water since he started physical therapy, demonstrating his own perception of meaningful change with aquatic physical therapy interventions. His current measured strength and ROM are provided in Tables 3 and 4, showing no change in either land-based assessment.

**Table 4.** Current Status Active Range of Motion (AROM)

	Right	Left
Hip Flexion	45*	60*
Knee Flexion	120*	90*

## Discussion

Although research is limited on the benefits specifically of aquatic therapy in individuals with Spinal Muscle Atrophy, a review reports on the benefits of aquatic therapy in a number of neurodegenerative disorders. The study examined a number of neurogenic disorders including Huntington's Disease, Parkinson's Disease, Multiple Sclerosis (MS), and Amyotrophic Lateral Sclerosis (ALS) and defined aquatic physical therapy as interventions performed in a warm, above 30°C, approximately 86°F, chest-deep pool using a variety of exercise modalities including aerobic, stretching, range of motion (ROM), resistance, and stability training tailored to the individual's abilities, which is in line with the interventions utilized for the patient in this case study. Functional gains and improved quality of life were demonstrated in all groups. The article reports that aquatic therapy has been especially beneficial in patients with MS due to reduced risk of falls or fear of falling while exercising, and significant improvements in pain intensity, spasticity, fatigue, disability, and autonomy of the patient all resulting in improved quality of life.<sup>6</sup> Another study reported that individuals with MS who participated in aquatic therapy had greater life satisfaction than those who did not participate in aquatherapy.<sup>6</sup> Aquatic therapy interventions were also found to increase Brain Derived Neurotrophic Factor (BDNF) levels, known to promote neuroplasticity and support improved CNS function, while land-based therapy in these patients did not.<sup>6</sup> While these studies reported positive outcomes with aquatic therapy for a number of diagnoses, other studies have found conflicting results. A number of observational studies as well as several retrospective studies that examined the impact of aquatic therapy on quality of gait and improvements in motor scale outcomes reported that some patients demonstrated improvements and some demonstrated no change.<sup>6</sup>

A 2016 Cochrane review on interventions effective for Type III SMA concluded that interventions should focus on strength and aerobic training. The review claims that the muscle training should aim to increase a person's functional performance, muscle strength, cardiopulmonary exercise capacity, and quality of life, and reduce their levels of fatigue.<sup>7</sup> Example exercises mentioned included cycling on an ergometer, running on a treadmill, and lifting weights. These types of exercise would be appropriate for an individual with the appropriate amount of volitional control to perform these tasks, such as individuals with type III SMA. In the case of a more severe form, such as the patient from this case study, an environment would need to be created to allow the individual to optimally utilize the muscular control he or she does have, such as in the pool. The aquatic environment decreases the effects of gravity through buoyancy, allowing for more movement. Additionally, it can create an environment of safety, enabling individuals to try more types of movement without the fear of falling.

This Cochrane Review also addresses the benefits of exercise in individuals with type III SMA. From a metabolic stand point, regular exercise has the ability to optimize resources and metabolic function in available muscle tissue as well as counteract further muscle deterioration that occurs secondary to inactivity. Functionally, strength training has the ability to improve performance of anti-gravity activities such as standing up, jumping, and stair climbing. Aerobic exercise training will enhance exercise capacity and also improve walking distance and endurance. Additionally, exercise may have a neuroprotective effect, which could be explained by a relationship between the maturation state of the motor unit and resistance to neuronal cell death. A preclinical study referenced in this review, studied SMA type II-like mice. The mice in this study participated in a running wheel training program showed an exercise-induced acceleration of the motor-unit maturation on the level of the motor neuron,

neuromuscular junction, and muscle fiber, and a delay in motor neuron death.<sup>7</sup> These findings would suggest the benefits of aerobic training in individuals type II SMA might be beneficial in slowing the progression of the disease process.

In this case study, the patient's available volitional control against gravity was significantly limited. However, in the water, the patient was able to move more continuously and with decreased assistance from the therapist, promoting increased use of volitional control. In the water, the patient was able to swim and performed modified walking with ankle weights, while on land the patient was limited to more static forms of exercises such as sitting on the edge of the table or working to maintain head control. The aquatic environment broadened the possibilities of movement with this patient, allowing for greater benefits with continuous movement and active range of motion potentially improving the patient's cardiovascular endurance and muscular endurance even if traditional manual muscle testing did not show any significant changes in muscular strength over the episode of care.

In addition to the benefits of utilizing an aquatic environment, it is also important to highlight alternative outcome measures that could discriminate the benefits associated with exercise, particularly when manual muscle testing and range of motion measurements do not reflect measurable or significant change despite patient impressions of improvement. A 2017 study aimed to identify perceptions of meaningful change in patients with primarily type II and non-ambulant type III SMA, concluded that meaningful change is relative to functional mobility abilities of the patient, avoiding decline in function, and that small changes, even when not reflected by an outcome measure, can be significant. The study took into account the perspective of the patient, the caregiver, as well as the clinician treating the child. The authors quote a parent of a non-ambulatory child diagnosed with type II SMA acknowledging the relativity of meaningful change, "I would be happy with crawling, but walking would be like hitting the lottery."<sup>8</sup> For the patient in this case study, walking and swimming in a pool such as the 17 year old in this case study was able to accomplish, would demonstrate progress with functional mobility, even if this was not reflected as measurable change on land. Additionally, the patient in this case study did not demonstrate a decline in function which can also be a form of meaningful change in this progressive disease.<sup>8</sup>

There is a SMA specific outcome measure known as the Hammersmith Functional Motor Scale (HFMSE)<sup>9</sup>. The outcome measure was designed to measure motor capabilities of non-ambulant SMA type II and ambulatory type III patients. There are 33 items on the HFMSE, which assess an individual's ability to perform actions such as sitting on a chair without support, rolling over from prone to supine position or the reverse, lifting the head from supine, getting up from lying, four-point kneeling, propping on arms, standing and stepping, kneeling, squatting, jumping, and walking up and down stairs.<sup>9</sup> The HFMSE was found to be a valid time-efficient outcome measure for clinical trials in SMA types II and III.<sup>9</sup> This outcome measure has been utilized as a baseline outcome measure for a recently approved drug called Spinraza.<sup>8</sup> However, patients report that this outcome measure is not sensitive to pick up small functional changes.<sup>8</sup> Partial improvement on an item is not accounted for on the HFMSE. Therefore, their total score would not detect improvements, while the patients would report that they have improved with certain skills or function. Similarly, the patient described in this case study would not have demonstrated change on the HFMSE if this outcome measure was utilized, as even though he, by clinician and patient observation, was moving more and able to hold positions for longer periods of time, highlighting limitations of this outcome measure.

Quality of life measures may be a way to measure additional benefits and change associated with utilizing aquatic therapy in individuals with limited volitional control, especially when gross motor and functional movement outcomes do not reflect meaningful change. In this case study, the patient was an adolescent, so the PedsQL would have been an appropriate outcome measure to assess changes in quality of life. The PedsQL accounts for physical, emotional and social function, as well as functioning at school. The PedsQL demonstrated responsiveness, construct validity, and predictive validity in hospitalized pediatric patients.<sup>10</sup> This may have the potential to translate into the outpatient pediatric setting, especially in children with neurodegenerative diseases. Because these individuals will likely not have rapid changes in their physical functioning score, improvements in their PedsQL score could be

reflective of improvements in emotional, social and school functioning scores, demonstrating improvements in participation of daily interactions and activities. The patient in this case study reported feeling like he could concentrate better in school because he was in less pain, for example, which would be reflected as an improvement in his PedsQL score, had we utilized this outcome measure throughout his care. Further research is needed to determine outcome measures that are sensitive enough to detect even small changes in volitional control or function in individuals with more severe degrees and types of SMA.

This case involving a 17 year old male with severe type II SMA highlights the perceived value of physical therapy, in particular aquatic exercise, to the patient. In these cases involving severe progressing disease, the individual's perception of change, even if it appears to involve only maintenance of function, may be a good indicator of the benefits of physical therapy interventions. Quality of life measures, such as the PedsQL in the pediatric population, also have the ability to detect social and emotional benefits of physical therapy in this patient population when gross motor gains are undetected by other outcome measures. Overall, physical therapy exercise interventions for individuals with SMA, as well as other degenerative neuromuscular diseases, are indicated for several reasons. The aquatic physical therapy environment can provide a safe environment that decreases the effects of gravity, allowing for greater autonomy with movement for individuals with very limited volitional control against gravity. This increased autonomy, as well as movement possibilities facilitated by the aquatic environment, can provide both functional as well as psychological benefits for individuals being treated with degenerative neuromuscular disorders. Lastly, for individuals with severe forms of SMA not able to participate in other forms of aerobic training such as cycling, walking, or running, aquatic exercise may be sufficient to induce neuroprotective adaptations. Thus, despite no improvements in ROM or strength, this case suggests that physical therapy consisting of land-based and aquatic-based interventions provided pain relief and possibly decreased the functional loss expected with this degenerative disorder over a two-year period. However, additional research is needed to fully determine the benefits of physical therapy interventions in this population.

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